Case Report

Persistent neutropenia presenting as recurrent infections in a child: A case report

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ABSTRACT

We are presenting a case of a 5-year-old girl with a history of recurrent infections from infancy. Every time, she had to be admitted and given intravenous antibiotics. While presenting to us, she had pallor, but other examination findings were apparently normal. In the complete blood count, there was severe neutropenia, which persisted on repeating the tests on several occasions. Her other tests did not show any significant results. After excluding major diseases in the workup, we started her on granulocyte colony-stimulating factor. She responded to it well. Sometimes, even after extensive evaluation, an attributable cause may not be found in a patient with persistent neutropenia. However, they respond well to conventional treatment.

Key words: Child patient, Neutropenia, Recurrent infection

Neutrophils act as a first line of defense of the innate immune system against infections in our body. An absolute neutrophil count (ANC) below the normal level for age and race is termed neutropenia. Neutropenia can be classified as acute or chronic and acquired or congenital [1]. Congenital neutropenia is a rare group of genetic disorders mainly caused by impaired neutrophil differentiation with maturation arrest at the promyelocyte stage [2,3]. Persistent low neutrophil levels in this condition predispose to recurrent infections. This condition is also associated with increased risk of developing myelodysplastic syndrome and leukemia [4].

Here, we are describing an interesting case of a child with persistent neutropenia with a history of recurrent infections and mouth ulcers, although common genetic mutations were not found.

CASE PRESENTATION

Our patient, a 5-year-old girl, presented with a history of recurrent fever and mouth ulcers (Fig. 1) since she was 7 months of age. She had a history of hospital admission and intravenous antibiotic use multiple times with diagnoses of diarrhea, acute respiratory infection, enteric fever, mastoiditis, cellulitis, etc. She is the only child born out of a non-consanguineous marriage. Her mother had a history of one stillbirth at 6 months of gestational age. There is no family history suggestive of inborn error of immunity or other major illness and infection. She is immunized as per age.

On general examination, pallor was present. Anthropometric examination showed weight for age between −1 and −2 standard deviation (SD), height for age between −2 and −3 SD, weight for height between −1 and −2 SD, and head circumference between −1 and −2 SD according to the World Health Organization growth charts. This suggests she had moderate stunting. Her systemic examination was normal.

Her initial complete blood count at our institute showed hemoglobin 9.8 g/dL, total leukocyte count 2730/µL with differential count of neutrophil 3.2%, lymphocyte 79.1%, monocyte 13.6%, eosinophil 3.7% with ANC 87/µL, platelet count 2.43 lakh/µL. Peripheral smear showed microcytic hypochromic anemia and leucopenia. Serial complete hemograms showed persistence of neutropenia, with most of the ANC readings below 500/uL (Table 1).

Hepatitis B, Hepatitis C, and human immunodeficiency virus (HIV) serology were negative; immunoglobulin profile was normal. T and B lymphocyte subset flow cytometry was normal. Dihydrorhodamine and nitroblue tetrazolium tests were negative, and antinuclear antibody profile was negative. Her vitamin B 12 and folic acid levels were in the normal range. Bone marrow aspiration

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Table 1: Some CBC values of our patient

Investigations	References	1st	2nd	3rd
Hb (g/dL)	11–14	9.8	10.2	10.1
TLC (/cu mm)	5000–15000	2730	3970	5140
DLC (%)	N-40–70 L-35–65 M-2–10 E-0–7	N3.2L79.1M13.6E3.7	N4.3L84M8.3E3.2	N1.5L85.9M6.9E5.4
ANC (/cu mm)	2000–7000	87	170	77
Platelet (lakh/cu mm	1.5–4	2.43	2.75	2.36

Hb: Hemoglobin, TLC: Total leucocyte count, DLC: Differential leucocyte count, ANC: Absolute neutrophil count, CBC: Complete blood count



Figure 1: Image showing a mouth ulcer

and biopsy showed normoblastic erythropoiesis, adequate and normal megakaryocytes, myelopoiesis with sequential maturation up to the neutrophil stage, and lymphopoiesis within normal limits. Genetic analysis (whole exome sequencing) suggested a heterozygous missense variant in Exon 5 (c428C>A) in the *SLC37A4* gene. A heterozygous mutation in Exon 28 (c.5897C>T) of the *ADGRV1* gene of uncertain significance was also detected. Her liver function tests, fasting blood sugar, lactate, uric acid, and lipid profile, including triglyceride levels in blood, were within the normal range.

She was started on granulocyte colony-stimulating factor (G-CSF) injection 5 µg/kg daily. Her neutrophil count gradually improved and normalized within days. As serial hemograms showed improvement in ANC readings, G-CSF was titrated to alternate days and subsequently to a twice-weekly regimen. A trial of any further decrease in G-CSF resulted in recurrence of neutropenia.

She is now growing well, maintaining normal blood counts on G-CSF. There was no history of hospital admission in the past 2 years.

DISCUSSION

Our patient presented with a past history of recurrent infections from an early age, for which she needed treatment with intravenous antibiotics and hospital admissions. There was persistent severe neutropenia, but most of the other tests were apparently normal. Neutropenia can be acquired or hereditary, acute or chronic [1]. In our patient, neutropenia persisted until she was given G-CSF. After attempting to stop G-CSF, there was a decline in ANC. Acquired neutropenia can be caused by infections, where neutropenia is usually transient, and some drugs. In our patient, there was no history of intake of any bone marrow-suppressing medications, and the viral markers for common viruses, such as HIV, were negative. Another important cause of acquired neutropenia is autoimmune neutropenia. Our patient's autoantibody test panel was negative. Primary autoimmune neutropenia is a rare cause of neutropenia. This disorder is associated with recurrent mild skin and upper respiratory tract infections [1].

Diagnosis is made by the presence of neutrophil-specific autoantibodies, but negative autoantibodies do not exclude the diagnosis. Bone marrow examination is normal [1]. We could not do the neutrophil-specific antibodies due to non-availability, and our patient had more severe infections than described in cases of primary autoimmune neutropenia.

Neutropenia is defined broadly as ANC <1500/µL which can further be subclassified as mild (ANC 1000–1500/µL), moderate (ANC 500–1000/µL), severe (ANC 200–500/µL), and very severe (ANC <200/µL). Our patient was falling in the very severe variant of neutropenia. Congenital neutropenia patients are prone to recurrent infections, such as otitis media, pneumonia, cellulitis, and osteomyelitis. A peculiarity of infections in these patients is the absence of pus [5]. Cyclical neutropenia, on the other hand, has a cyclically low neutrophil count over a 21-day period. Some mutations of congenital neutropenia are ELA2, HAX1, GFI1, WAS, SLC37A4, and G6PC3 [5]. Genetic study of our patient showed a heterozygous mutation in the gene *SLC37A4*. Mutation in the *SLC37A4* gene has been associated with glycogen storage disease type Ib, caused by a mutation in the transporter for glucose-6-phosphate. This disease is inherited in an autosomal recessive pattern, characterized by hypoglycemia, high fasting lactate levels, and glycogen overload in the liver and congenital neutropenia. There is no maturational arrest of neutrophils in this type of congenital neutropenia [6]. However, in our patient, the mutation was heterozygous and reported as a variant of unknown significance. With the available resources, we could not find out a cause for our patient. The genetic basis of severe congenital neutropenia remains unknown in a significant proportion of children [7].

Recombinant human G-CSF is the first-line of therapy in congenital neutropenia. It is given at a dose of 3–5 µg/kg/day subcutaneously. It stimulates the bone marrow to produce and release neutrophils into the circulation, hence significantly reducing the severity of infections [5].

CONCLUSION

Sometimes even after extensive evaluation, an attributable cause may not be found in a patient with persistent neutropenia. But they respond well to conventional treatment.

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